

What is CAH?

Congenital adrenal hyperplasia (CAH) is a disorder in which the adrenal glands (located above the kidneys) do not produce enough of the adrenal hormones cortisol and aldosterone. Cortisol ensures an adequate energy supply and blood sugar level and helps to maintain normal amounts of sodium and potassium, which are important for normal cell functioning and in the regulation of body fluids. When the body does not produce enough of these hormones, symptoms can occur: low blood sugar, drowsiness, loss of consciousness, coma, vomiting dehydration, extreme muscle weakness, weakness of the heart, and poor growth.

What causes CAH?

CAH is inherited from both parents. It cannot be “caught” like a cold or “given” to another child.

How is CAH found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Services’ Inborn Errors of Metabolism Laboratory, where it is tested for congenital adrenal hyperplasia and several other conditions. These tests are all part of the State’s Newborn Screening Program. If the results are not within an acceptable range, the baby’s doctor and parents are notified.

What does the lab look for?

The lab tests for a hormone in the blood, 17-alpha-hydroxyprogesterone (17-OHP). This hormone is higher in babies who have CAH.

Does an abnormal screening test mean that my baby has CAH?

Screening tests always need to be confirmed by additional testing and medical evaluation.

What will my baby’s doctor do?

Depending on the results of the screening, your doctor may repeat the testing or refer your baby to a specialist (pediatric endocrinologist). Your doctor and the specialist will confirm the diagnosis by doing one or more of the following:

- Getting information about your baby
- Examining your baby
- Taking a blood sample to confirm the screening results

How is CAH treated?

CAH is a lifelong, but treatable medical condition. Without treatment, a baby with CAH is at risk for complications. Treatment for CAH is by taking daily hormone replacement medications by mouth. These medications help the body to maintain a normal salt balance (preventing dehydration), to maintain a normal blood sugar (providing energy), and to promote normal growth and normal puberty. Medication is taken for life and doses are adjusted when necessary. Children with CAH need to be followed closely to monitor growth.

**IF YOU ARE ASKED TO HAVE
YOUR BABY RE-TESTED, ACT
QUICKLY AND FOLLOW YOUR
DOCTOR’S ADVICE**

For more information, please contact:
The New Jersey Department of Health
and Senior Services

Newborn Screening and Genetic
Services

at (609) 292-1582

or

The Inborn Errors of Metabolism
Laboratory at (609) 292-3090

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Congenital Adrenal Hyperplasia (CAH)

Important Information for
Parents



